

# **SATELLITE MEETING**

## **EUROPEAN NETWORK FOR LAMINOPATHIES MEETING**

**September 3-5 2019**

**Bush House, Kings College London**

**London, UK**



# EUROPEAN NETWORK FOR LAMINOPATHIES MEETING

## Programme

September 3 2019	morning	Time
<b>DIAGNOSIS, FOLLOW-UP, NEW THERAPEUTIC TOOLS</b> Chairs: Giulia Ricci and Karima Djabali		11:30 - 13:00
Common mechanism: <b>Does a point mutation alter the structure of a lamin filament?</b> Ohad Medalia - Switzerland		11:30 - 11:45
Muscular Laminopathies: <b>Deflazacort treatment in LMNA-related congenital muscular dystrophy: design of an ongoing Italian cohort pilot study.</b> Giulia Ricci - Italy		11:45 - 12:00
Progeroid Laminopathies: <b>Autophagic Removal of Farnesylated Carboxy-Terminal Lamin Peptides</b> Karima Djabali - Germany		12:00 - 12:15
Progeroid Laminopathies: <b>First-in-the-world: TAVI in a patient affected by Hutchinson-Gilford Progeria syndrome</b> Francesco Musumeci - Italy		12:15 - 12:30
Lipodystrophic laminopathies: <b>Type 2 Familial Partial Lipodystrophy (Dunnigan's Disease): Allelic heterogeneity and variable expressivity</b> David Araujo-Vilar - Spain		12:30 - 12:45
<i>Short talk Henner Kalden</i> <b>AEGERION Novelion</b>		12:50 - 13:00
<b>LUNCH</b>		13:00 - 14:00

September 4 2019

morning

Time

**PATIENT ASSOCIATIONS, PATIENT CARE, NEW THERAPEUTIC TOOLS**

Chairs: Lorenzo Maggi and Marjet Stamsnijder

11:30 - 13:15

**Muscular Laminopathies: Natural history and clinical trial readiness in striated muscle laminopathies- the ENMC workshop in 2020**

Lorenzo Maggi - Italy

11:30 - 11:45

**Muscular Laminopathies: Muscular Laminopathies – Patients Association speeches A.I.D.M.E.D Onlus and Associazione Alessandra Proietti Onlus**

Stefania Fazioli, AIDMED and Irene Proietti, Associazione Alessandra Proietti - Italy

11:45 - 11:55

**Muscular Laminopathies: EDMD patient perspectives**

Salomè Mist Kristiansdottir and Eleonora Cugudda - EDMD Facebook Group

11:55 - 12:10

**Cardio-laminopathies: Cardio-laminopathies, recent updates**

Andrea Barison and Giovanni Peretto - Italy

12:10 - 12:25

**Cardio-laminopathies: An AAV delivered therapeutic that suppresses Lamin-induced Dilated Cardiomyopathy**

Colin Stewart - Singapore

12:25 - 12:40

**Lipodystrophic Laminopathies: AELIP: Multidisciplinary intervention from a socio-sanitary scope, to improve the quality of life of people and relatives affected by lipodystrophy**

Naca Eulalia, AELip - Spain

12:40 - 12:50

**Progeroid laminopathies: The importance of Peer Groups**

Marjet Stamsnijder, Progeria Family Circle - EU

12:50 - 13:00

**Progeroid Laminopathies: A patient's perspective about the research: from scientific disclosure to the lab**

Sammy Basso, AIProSaB - Italy

13:00 - 13:15

**LUNCH**

13:15 - 14:15

September 4 2019

afternoon

Time

## **European Network for Laminopathies management**

Chairs: Gisèle Bonne and Giovanna Lattanzi

18:15 - 19:15

### **Network organization**

#### **The European Network for Laminopathies as a legal entity**

Giovanna Lattanzi - Italy

18:15 - 18:30

### **Network activities**

Gisèle Bonne

18:30 - 18:45

### **Network relationship with ECLip and other consortia**

#### **The European Consortium of Lipodystrophies (ECLip)**

David Araujo-Vilar - Spain

18:45 - 19:00

### **Proposals and discussion**

All partners

19:00 - 19:15

September 5 2019

morning

Time

## **PATHOMECHANISMS AND THERAPIES**

Chairs: Pavel Hozak and Nolwenn Briand

11:30 - 13:00

Common mechanisms: **Inositol phosphates functions in regulation of gene transcription**

Pavel Hozak - Czech Republic

11:30 - 11:45

Progeroid Laminopathies: **Endothelial progerin expression causes cardiovascular pathology through a paracrine profibrotic signaling**

Roland Foisner - Austria

11:45 - 12:00

Muscular and Progeroid Laminopathies: **Laminopathy discoveries in Sweden** Einar Hallberg - Sweden

12:00 - 12:15

Lipodystrophic laminopathies: **Lipodystrophy-causing lamin A mutations remodels local and global chromatin architecture**

Nolwenn Briand, Norway

12:15 - 12:30

Cardiolaminopathies: **Haploinsufficiency and dominant negative effects of mutant lamin A/C both contribute to the increased severity of L-CMD compared with EDMD**

Anne Bertrand, France

12:30 - 12:45

**Conclusions**

12:45 - 13:00

**LUNCH**

13:00 - 14:30

**END OF ENL MEETING**



FUNDACIÓN  
**ANDRÉS MARCIO**  
niños **contra** la laminopatía



Alessandra  
Proietti  
O.N.L.U.S.



Associazione Italiana Distrofia  
Muscolare di Emery Dreifuss (ONLUS)



**cells**



**cost**  
EUROPEAN COOPERATION  
IN SCIENCE & TECHNOLOGY



EuroCellNet



**SARSTEDT**

